

(FILE 'HOME' ENTERED AT 17:12:50 ON 22 NOV 2002)

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:12:59 ON 22 NOV 2002

L1 49 S CD38 (P) GENE (P) SEQUENCE  
L2 30 DUP REM L1 (19 DUPLICATES REMOVED)

FILE 'STNGUIDE' ENTERED AT 17:20:25 ON 22 NOV 2002

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:25:42 ON 22 NOV 2002

L3 10 S CD38 AND (DIABETES OR IDDM OR NIDDM) AND (MUTATION? OR POLYMO  
L4 7 DUP REM L3 (3 DUPLICATES REMOVED)  
L5 6 S L4 NOT L2

FILE 'STNGUIDE' ENTERED AT 17:27:33 ON 22 NOV 2002

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:34:56 ON 22 NOV 2002

L6 17 S CD38 AND INSULIN AND (MUTATION? OR POLYMORPHISM?)  
L7 13 DUP REM L6 (4 DUPLICATES REMOVED)  
L8 6 S L7 NOT L4

FILE 'STNGUIDE' ENTERED AT 17:37:12 ON 22 NOV 2002

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:39:27 ON 22 NOV 2002

L9 267 S (MODY1 OR MODY2 OR MODY3) AND (MUTATION? OR POLYMORPHISM?)  
L10 171 DUP REM L9 (96 DUPLICATES REMOVED)  
L11 6 S L10 AND TYPE 1 AND TYPE 2

L11 ANSWER 3 OF 6 MEDLINE  
 AN 2001322865 MEDLINE  
 DN 21202573 PubMed ID: 11307309  
 TI Diabetes mellitus.  
 AU Iwasaki N  
 CS Diabetes Center, Tokyo Women's Medical University, Shinjyuku-ku, Tokyo  
 162-8666.  
 SO RINSHO BYORI. JAPANESE JOURNAL OF CLINICAL PATHOLOGY, (2001 Feb) 49 (2)  
 161-4. Ref: 0  
 Journal code: 2984781R. ISSN: 0047-1860.  
 CY Japan  
 DT Journal; Article; (JOURNAL ARTICLE)  
 General Review; (REVIEW)  
 (REVIEW LITERATURE)  
 LA Japanese  
 FS Priority Journals  
 EM 200106  
 ED Entered STN: 20010611  
 Last Updated on STN: 20010611  
 Entered Medline: 20010607  
 AB Diabetes mellitus is a group of metabolic disorders characterized by  
 hyperglycemia resulting from defects in insulin secretion, insulin action  
 or both. Genetic factors contribute to the development of diabetes. Some  
 forms such as the condition called maturity-onset diabetes of the  
 young(MODY) result from **mutations** in a single gene. Other forms  
 such as **type 1** or **type 2** diabetes  
 are multifactorial in origin with different combinations of genes together  
 with non-genetic factors contributing to the development of hyperglycemia.  
 MODY has been a good model for studying the genetics and pathophysiology  
 of diabetes. This form of diabetes can result from **mutations** in  
 at least seven different genes: hepatocyte nuclear factor(HNF)-4 alpha/  
**MODY1**, glucokinase/**MODY2**, HNF-1 alpha/**MODY3**,  
 insulin promoter factor(IPF-1)/**MODY4**, HNF-1 beta/**MODY5**, NeuroD1/**MODY6** and  
 Islet(Isl)-1/**MODY7**. **Mutations** in HNF-1 alpha/**MODY3** are  
 the most common cause of MODY in Japanese identified to date accounting  
 for about 15% of cases of MODY. **Mutations** in the HNF-4 alpha/  
**MODY1**, glucokinase/**MODY2**, HNF-1 beta/**MODY5** and  
 Isl-1/**MODY7** genes have also been found in Japanese; however, they are rare  
 causes of MODY. Clinical studies indicate that patients with MODY are  
 generally not obese and that all forms of MODY are characterized by  
 pancreatic beta-cell dysfunction. Patients who have **mutations** in  
 the HNF-1 beta/**MODY5** gene have non-diabetic kidney dysfunction including  
 renal cysts. Female carriers may also exhibit abnormalities in the upper  
 vagina and uterus. Genetic approach for **type 2**  
 diabetes had done by using non-parameteric linkage analysis such as  
 sibpair analysis which worked well and NIDDM1 and NIDDM2 have been  
 identified to date. The responsible gene for NIDDM1 was recently  
 identified to be Calpain 10, and SNP43 in this gene could explain all of  
 the evidence for linkage in Mexican American **type 2**  
 diabetes.

L2 ANSWER 10 OF 30 CAPLUS COPYRIGHT 2002 ACS  
 AN 2000:815089 CAPLUS  
 DN 133:359765  
 TI Diabetes risk factor detection with CD38 gene mutations  
 IN Egashira, Toru; Nagano, Makoto; Sagehashi, Yukiko; Matsui, Kana; Hattori,  
 Hiroaki; Kanazuka, Akira; Takasawa, Noboru; Okamoto, Hiroshi  
 PA BML Inc., Japan  
 SO Jpn. Kokai Tokkyo Koho, 19 pp.  
 CODEN: JKXXAF  
 DT Patent  
 LA Japanese  
 FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	JP 2000316578	A2	20001121	JP 1999-131955	19990512
AB	Assessment of diabetes risk factors by detecting mutations in gene coding for type II transmembrane glycoprotein CD38 (ADP-ribosyl cyclase/cyclic ADP-ribose hydrolase), is disclosed. Polymorphism was obsd. in the exon 3 and 4 of CD38 gene from peripheral leukocytes; the corresponding point mutation of exon 3 show R140W missense mutation and that of exon 4 showed silent mutation at I 168. The allele frequency of R140W missense mutation was different between NIDDM and non-diabetic controls. Glut 2 gene mutation was also obsd. in a family with the CD38 gene mutation, indicating the involvement of CD38 gene and other genes in pathogenesis of NIDDM. DEEG (denaturing gradient gel electrophoresis), direct sequencing, and PCR-RFLP were used.				